FATTY ACID OXIDATION DISORDERS

What is it?

Fatty acid oxidation disorders are a group of inherited metabolic conditions that lead to an accumulation of fatty acids, and a decrease in cell energy metabolism. Each fatty acid oxidation disorder is associated with a specific enzyme defect in the fatty acid metabolic pathway and affects utilization of dietary and stored fat.

How do you get it?

All fatty acid oxidation disorders are inherited in an autosomal recessive pattern. As an autosomal recessive disorder, the parents of a child with one of these conditions are unaffected, healthy carriers of the condition, and have one normal gene and one abnormal gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with two copies of the abnormal gene and the resulting organic and acid defect. Carrier parents have a 50 percent chance of having an unaffected, non-carrier child. These risks would hold true of reach pregnancy. All siblings of infants diagnosed with a fatty acid organic disorder should be tested.

Affected infants can be diagnosed in the neonatal period. These disorders can cause recurrent episodes of hypoglycemia; clinical findings may include lethargy, hypotonia, failure to thrive, persistent vomiting, hepatomegaly, rhabdomyolosis and Reye syndrome-like episodes.

How common is it?

Medium chain acyl-CoA dehydrogenase deficiency (MCAD) is the most common of the fatty acid oxidation disorders with an incidence of approximately 1 in 10,000 – 50,000 births.

The precise incidence of other fatty acid oxidation disorders is unknown; however, these are rare conditions with an estimated incidence of 1 in 100,000 births or less.

How is it treated?

Early diagnosis and treatment is essential for an improved prognosis. If left untreated these conditions may result insignificant disability and ultimately, death. Most of these conditions are chronic, with life-long episodes of hypoglycemia. In some of the more severe infantile forms, there is a very poor prognosis. For most fatty acid oxidation disorders, management

involves long-term monitoring of serum glucose, a low-fat/high-carbohydrate diet, supplemental carnitine and avoidance of fasting. Aggressive medical management is necessary during illness, especially if your child is vomiting or is not receiving adequate nutritional intake. At the time of inter-current illness, your infant/child should be admitted for medical care, including administration of intra-venous dextrose to prevent hypoglycemia. If your child needs additional testing or diagnostic evaluation, it is important that you follow through and obtain this. Treatment is life long and compliance with dietary management is imperative to your child's health, growth and development.

Infants and children with a fatty acid oxidation disorder should have regular follow-up appointments with a metabolic disease specialist. If your infant shows early signs of the condition, such as vomiting or lethargy, immediately seek medical care. A medical plan developed by the metabolic specialist and the primary care provider should be developed for these acute episodes.

Long-term management, monitoring and compliance with treatment recommendations are essential to your child's well being. A multi-disciplinary approach including the following specialties is recommended: pediatrics, genetics and nutrition.

Where can I get services?

Cardinal Glennon Memorial Hospital for Children
St. Louis, MO
314-577-5639

Children's Mercy Hospital
Kansas City, MO
816-234-3804

St. Louis Children's Hospital
St. Louis, MO
314-454-6051

University Hospital and Clinics
Columbia, MO
573-882-6979

Related Links

Centers for Disease Control and Prevention www.cdc.gov

Medline Plus (National Library of Medicine and the National Institutes of Health) www.medlineplus.gov

National Library of Medicine (NLM) Pub Med www.ncbi.nlm.nih.gov

The Fatty Oxidation Disorders (FOD) Family Support Group
www.fodsupport.org